



SEQUENCE LISTING

(1) GENERAL INFORMATION:

- (i) APPLICANT: Ruddy, David A.
Wolff, Roger K.
- (ii) TITLE OF INVENTION: POLYMORPHISMS IN THE REGION OF THE HUMAN
HEMOCHROMATOSIS GENE
- (iii) NUMBER OF SEQUENCES: 24
- (iv) CORRESPONDENCE ADDRESS:
 - (A) ADDRESSEE: TOWNSEND and TOWNSEND and CREW LLP
 - (B) STREET: Two Embarcadero Center, 8th Floor
 - (C) CITY: San Francisco
 - (D) STATE: CA
 - (E) COUNTRY: USA
 - (F) ZIP: 94111
- (v) COMPUTER READABLE FORM:
 - (A) MEDIUM TYPE: Floppy disk
 - (B) COMPUTER: IBM PC compatible
 - (C) OPERATING SYSTEM: PC-DOS/MS-DOS
 - (D) SOFTWARE: PatentIn Release #1.0, Version #1.30
- (vi) CURRENT APPLICATION DATA:
 - (A) APPLICATION NUMBER: US 08/852,495
 - (B) FILING DATE: 07-MAY-1997
 - (C) CLASSIFICATION: 435
- (vii) PRIOR APPLICATION DATA:
 - (A) APPLICATION NUMBER: US 08/724,394
 - (B) FILING DATE: 01-OCT-1996
 - (C) CLASSIFICATION:
- (vii) PRIOR APPLICATION DATA:
 - (A) APPLICATION NUMBER: US 08/652,265
 - (B) FILING DATE: 23-MAY-1996
 - (C) CLASSIFICATION:
- (vii) PRIOR APPLICATION DATA:
 - (A) APPLICATION NUMBER: US 08/630,912
 - (B) FILING DATE: 04-APR-1996
 - (C) CLASSIFICATION:

(2) INFORMATION FOR SEQ ID NO:1:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 235033 base pairs
 - (B) TYPE: nucleic acid

(C) STRANDEDNESS: single

(D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:1:

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CTATTTTAGA TAGCCTTGTC TGAAACAGAG CTGGGACCTG ATGAGTGAAA ATGAGCTCAC	180
CAGAAGAAAA ATCAAACAGG CATTTTCAGAG ATTGAGGCCA AGAAGTTAAA TGTCTTAAAT	240
GGGCAGAGCT TAGCTGCTTG ATGTGAAAAG AGACCAGCGT GGCTGGAACA GCAAAGGAGA	300
ACAGCAGAAG AGGTGAACAG AGGCCAGAGA TGGTCACTGA GTGGGCCCTT AAGTCATGGT	360
AAGGAGTATG GAGAATGAAT TATTGCATGT ATTGAATATG TAGGTGACGT GACTCACAGA	420
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GAGTAAATGG TAGTGTCAGT TATTGAACTG GGGAGAACTG GAAGGGATAA CAGGCTTAAG	540
GAGCACGTTT ATTCTGTGT CTTGGAAGTG TTTAGGGTGA AAGACCTATT AGAGTTCTAA	600
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CCATCTGTTA	GCCAGGAACA	GATTCTCACC	ATAAACTATG	TTGGCACCTT	GATCTTAAAC	4200
TTCCAGGCTC	CAAACTGTG	AGAAAATGAA	TTTCTGTTCC	AAGCCTCTTA	GATATGGAAA	4260
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CATCCAATGA	AGTTCTGACA	TTTCTTCAAC	ATGAGTACAG	TAATTCAATG	CCAGAGAATT	4560

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CAAACTGCCA	GTGAAATTTG	GATTTTTGGA	ATATAGTTTC	TTTTTTCTTG	TTACTTTTTG	6060
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GGTGAACACC	GCCACACCTG	ACTAATTTGT	GTAGTTTTAT	TAGAGATGGG	GTTTCGCCAT	6420
GTTGGCCAGG	CTGGTCTCAA	ACTCCTGACC	TCAGGTGATC	TACCCACCTC	AGCCTCCCCA	6480
AGTGCTGGGA	TTACAGATGT	GAGACACCAG	ATCAGCCTCA	GAAGACATTT	TCTATTGGAA	6540
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AATAAACCAA	CTCTCTACAA	CAAAGTGCTT	CCTGGCTGCC	TAAGTCATTG	ATTCATTCAG	6660
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GATAAACCT	ATCTCTTAGA	TTGTTGGATT	AAATGAATTA	ACATACTGGA	AGCTCATGAA	7380
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CTGGATCCGC	CCTGAGCCGG	TGTCAGTATC	TGGGAAGTGG	GAGGCGCGTC	AGCAGTAAAC	7980
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CACCACGCCC	AACTTATTTT	TTTATTTATT	ATTTTTATTT	AGTAGAGAGG	TGTTTCACCA	8820
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AGTGGTAGGA	TTAGAGGGGT	GAGCAGAAAG	CAAAGGTTTT	TGAGTGGCCA	CAGGCCCCAC	8940
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CGGGGGACTA	GTCGGAGGAC	CAAACAAGGT	TACCAACACG	TTAGAGTTTT	GCCTTCAATT	9240
TACATTTTTA	AAGTAATCAC	AACGAAGTGT	TTAGATCACG	AGGCATCCCT	GCATGTAAAC	9300
TGTTAGGCAC	TAACTATGGT	CGATCTTACA	AAGCATTAAC	TAGAATATTT	CTTTAGAGTA	9360
TGATAGTACG	TAACTGACCT	ACTATTACAT	ACAAACAGAC	CAACCTTTAG	TAACAGCGCT	9420

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GCCTTCGAAC	ACTGAACTGA	AGGCCAGTAA	GGACTAGGCG	CTGGGTGGGG	GAGAATGAAG	11640
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GCTCACTCAG	AAGAAACACA	ATGAGACAAG	AGAAGAGCCA	TCTCCTTCCT	TCTCTATTTA	38580

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CATGCCATTC	TCCTGCCTCA	GCCTTCCGAG	TAGCTGGGAC	TACAGGCGTC	TGCCACCACG	38880
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CTTCAAGGGA	TCCTCCTGCC	TTGGTCTCAC	AAAATGCTGG	GATGACAGAC	ATGAGACACC	225480
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TAGTTAGCCA	GCTACTTTTT	GTAGGAGAGC	TATGTTTAGG	CTAGGTGCTA	TAGGATTCTC	226200
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GATTCCCAAA	CACGGTCTTT	TCATATACAT	TTTCCACTGT	ACATACTTTC	TGACCTGGAA	231060
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(2) INFORMATION FOR SEQ ID NO:2:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 237326 base pairs
- (B) TYPE: nucleic acid
- (C) STRANDEDNESS: single
- (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:2:

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CTATGTTGGC	CAGGCTAGTT	TGGAATCCT	GACCTCCAGT	GATCCATTCT	CATTGGCCTC	2940
CCAAAGTGCT	GGGATTACAG	GCAGGAGCCG	CCAGGGCTGC	CACTTTGATG	TCAGACTCAG	3000
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TCAAAGATGC	CCTGCAGAAC	TGTGTGGGAG	TCTCTCACAG	ATGGCTGCCT	GGGTGGGACC	3120
CCACCAAACCT	GAAAGACCGA	GACTTCAGGC	AGGGCAGATG	GAGTAGGCCA	ACTACAGAGC	3180
CAGAGGTGAC	ACTGAGACAC	CACTGGGCCT	GGAAATCAGG	GCATCAAGCC	AAAGAGGGTT	3240
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CATGTCTCAG	CCTCCAGAGT	AGCTGGGACT	ACAGGCGCCC	GCCACCACGC	CTGGCTAATT	3720
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CATGCCCCGC	TGGGATGGAA	TAAATTTATC	TTGTATGGGA	GAAGGACATA	CATTTTGGCA	3900

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ATTAAAAGAC	CTCCAGAATT	CTGGAAGTAG	GAAGTTTCCT	CTTCAAAGTC	TACAGAGGAA	15540
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CCTTTTCTCC	ATTATCTGTC	TTTCCAGTGA	TGAAATTTTG	ATCTGGCCCT	CCCAAGTATT	15660
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GCTCTTCCCC	AGGGGGTACA	GCCAAGGTTA	TCCAGCCCTG	GTAGGTCCCA	TCCCCATTGG	41340
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AAGCTTATGT	TTGCTTAATG	CTTGGTAAAA	TTCCTGGTAC	ATGGTAACCA	CCTAATAAGT	153940
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GTCAAAGGAA	TGAGAAAAGA	CAAGTTAAGA	GTGCATAAAG	TGGGTCCAGG	GTGCCAGCAC	154060
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ACAAAGAAGC	AGGTGGTGAG	GACGTGAGGG	TAAACAGGTG	AGGGCATGAG	GACATGGGGG	154180
TAGAAAGGTA	GTGGTGCATT	AAGCGTAGCT	GTGACAGTTT	AGCATTTTCT	TTGACACATG	154240
TAGAATATAC	TCTGCTGCTT	GAGATAGTAG	AGGACACGTT	TATGAGTGAA	AAGCAAGGAA	154300
CCAACAAGTC	TGTGCACTTT	CCAGAGGCTA	TGAGGGGTTT	TATGCCCTGA	GCCCTGGGTT	154360
CCATCCAAGC	CACAAGGGGT	TTTATGCCCT	AGGCTTAGAT	TTGTGGTGCG	GCAGGGCAGC	154420
CTTCCACCAT	TTGGCACAGA	GCTTGGTGTT	CCAAAGGCCA	CGAGGGGTTT	TGGACCCTGG	154480
ACCCCGGACA	TCTTCCAAGA	CTCTTTTACA	TTATGACAGA	CAAGCCAGTC	CTGCTTCAGC	154540
TCTTCTAACA	ACATGTAGTA	ATAATGATAT	CATCAACATC	ATCTTCGTCT	TAATTATTCA	154600
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ACTTTTTCTA	CCAGAACTGG	TATCAGCTCA	TGCTCTGCCT	TATGCAAATT	AAGAAAATAT	203940
CATACCTTTT	GGGTAAATTA	AGCCAAGAAA	GTTCTCCTTT	CTTCTCTTTC	TCTCTTTCTT	204000
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TGCTAATGAT	TAACACATTC	ACCTTGGCTC	TTGGTTTGCC	TGCTCCCTCT	TCTTTTATCT	229020
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AGAGGACTGG	CCAGAGTGGG	AATGTTCTGA	ATTCAGAATA	ACTGAAGCAG	TACAGGATAG	229140
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AAGAAATCTT	GGAAATAGGT	TTATATAAAA	TTTATTTTTT	CCTTTTTTAT	TGTTATGGAA	230760
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GGGACTCCTC	TTTGTAGCTC	CAAGTGCCAC	TAACAATTCT	TAGGACCTGA	GCTATAAGCC	230880
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GAAGATGATT	CTGCCTCTTT	TAATATGTGA	CTCCTCAGAT	TCAGAAAGTG	CTCGCTAGTC	231120
TTAAGAGTGA	ATTACCCTCA	GTGGTCCAGC	GCTTATGAAC	CCACATCTAA	CCCTATCCCC	231180
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CGGAGTGCTG	AGAGCCTTCT	TAGTGTGTAT	TCAGTGTTTT	AAGAGAGCTT	GTGGATGAAT	232260
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TGTTCCCAGG	AAGAATCAGT	CACCCAACAG	TGTCCTTGTC	ACATCTTAGG	TTCTACACCT	233460
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CCACCTCCTG	GGTTCAAGTG	ATTCTCCTGC	CTCAGCCTCC	TGAGTAGCTG	GGATTACAGA	233640
CGTGTGTCAC	CACACCTGGC	TAATTTTTGT	ATTTTGTAGT	GAGACAGGGT	TTCACCGTGT	233700
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CATTGGGGCC	AAATCTTAGA	TCATGTAAAT	TTTCTTCTAT	ATTTTATTCT	AAAAGCTTGT	235920
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CTCATCTGGT	CTCCTGCTCT	TGAACTGGGA	TTTACATCAT	CAGTTCCTCT	GGTTCTCAGG	236520
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ATTTAGGAAT	TTGCCTTACC	AAGTAAAACA	TAAGGGCAGC	TGAGGTGCTG	ACTGAAGACA	237180

AATGGAGCAT AGAATAAGAG TAGTAAAGAA TGCCAAAAAT GCTGTCATGT ATCCATTGAC 237240
AAAAGGAGCT ATAAAGCCTT TAGGTATTTT CACACTTGCT CTGTTACGTA AATGTATGTG 237300
TGTGTGTGTG TGTGTGTGTG TGTGTG 237326

(2) INFORMATION FOR SEQ ID NO:3:

- (i) SEQUENCE CHARACTERISTICS:
(A) LENGTH: 20 base pairs
(B) TYPE: nucleic acid
(C) STRANDEDNESS: single
(D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:3:

CGTTAGAACG CGGCTACAAT 20

(2) INFORMATION FOR SEQ ID NO:4:

- (i) SEQUENCE CHARACTERISTICS:
(A) LENGTH: 20 base pairs
(B) TYPE: nucleic acid
(C) STRANDEDNESS: single
(D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:4:

GCCGATTCAT TAATGCAGGT 20

(2) INFORMATION FOR SEQ ID NO:5:

- (i) SEQUENCE CHARACTERISTICS:
(A) LENGTH: 20 base pairs
(B) TYPE: nucleic acid
(C) STRANDEDNESS: single
(D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:5:

CTGTAAAACG ACGGCCAGTC

20

(2) INFORMATION FOR SEQ ID NO:6:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 19 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:6:

GCAGGAAACA GCTATGACC

19

(2) INFORMATION FOR SEQ ID NO:7:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 21 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:7:

GCATCAGCGA TTAACCTCTA C

21

(2) INFORMATION FOR SEQ ID NO:8:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 22 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:8:

TTGCATTGTG GTGAAATCAG GG

22

(2) INFORMATION FOR SEQ ID NO:9:

- (i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 21 base pairs
- (B) TYPE: nucleic acid
- (C) STRANDEDNESS: single
- (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:9:

CTGAGTAATT GTTTAAGGTG C

21

(2) INFORMATION FOR SEQ ID NO:10:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 21 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:10:

CTGAGTAATT GTTTAAGGTG T

21

(2) INFORMATION FOR SEQ ID NO:11:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 20 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:11:

AGAAGAGATA GATATGGTGG

20

(2) INFORMATION FOR SEQ ID NO:12:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 20 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:12:

GAATGTGACC GTCCCATGAG

20

(2) INFORMATION FOR SEQ ID NO:13:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 28 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:13:

CAACTGAATA TGCAGAAAAA AGTACACC

28

(2) INFORMATION FOR SEQ ID NO:14:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 21 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:14:

AGTAGCTGGG ACTCACGGTG T

21

(2) INFORMATION FOR SEQ ID NO:15:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 21 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:15:

AGTAGCTGGG ACTCACGGTG C

21

(2) INFORMATION FOR SEQ ID NO:16:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 21 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:16:

GCGCCACCAC TCCCAGCTCA T

21

(2) INFORMATION FOR SEQ ID NO:17:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 14 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:17:

TATATATATA TATA

14

(2) INFORMATION FOR SEQ ID NO:18:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 14 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:18:

AAAAAAAAAA AAAA

14

(2) INFORMATION FOR SEQ ID NO:19:

- (i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 10 base pairs
- (B) TYPE: nucleic acid
- (C) STRANDEDNESS: single
- (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:19:

AGGATGGTCT

10

(2) INFORMATION FOR SEQ ID NO:20:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 12 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:20:

TTGTTGTTGT TG

12

(2) INFORMATION FOR SEQ ID NO:21:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 14 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:21:

CTTTTTTTTT TTTT

14

(2) INFORMATION FOR SEQ ID NO:22:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 12 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:22:

AATAATAATA AT

12

(2) INFORMATION FOR SEQ ID NO:23:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 11 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:23:

TTTTTTTTTTT T

11

(2) INFORMATION FOR SEQ ID NO:24:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 20 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:24:

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